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"I have fought for so many things": Disadvantaged families' efforts to obtain community-based services for their child after genomic sequencing

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Abstract

Background: Families whose child has unexplained intellectual or developmental differences often hope that a genetic diagnosis will lower barriers to community-based therapeutic and support services. However, there is little known about efforts to mobilize genetic information outside the clinic or how socioeconomic disadvantage shapes and constrains outcomes.

Methods: We conducted an ethnographic study with predominantly socioeconomically disadvantaged families enrolled in a multi-year genomics research study, including clinic observations and in-depth interviews in English and Spanish at multiple time points. Coding and thematic development were used to collaboratively interpret fieldnotes and transcripts.

Results: Thirty-two families participated. Themes included familial expectations that a genetic diagnosis could be translated into information, understanding, and assistance to improve the quality of a child's day-to-day life. After sequencing, however, genetic information was not readily converted into improved access to services beyond the clinic, with families often struggling to use a genetic diagnosis to advocate for their child.

Conclusion: Families' ability to use a genetic diagnosis as an effective advocacy tool beyond the clinic was limited by the knowledge and resources available to them, and by the eligibility criteria used by therapeutic service providers' – which focused on clinical diagnosis and functional criteria

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more than etiologic information. All families undertaking genomic testing, particularly those who are disadvantaged, need additional support to understand the limits and potential benefits of genetic information beyond the clinic.

Introduction

Families of children with unexplained intellectual or developmental differences and complex health needs shoulder significant caregiver responsibilities, including seeking and securing therapeutic and support services. Service needs may include medical care, assistive technologies and educational and therapeutic services, e.g. special education, occupational therapy, and speech therapy (Henderson, Skelton, and Rosenbaum 2008; Newacheck et al. 1998). Parents may also require in-home supportive services or other forms of respite care to alleviate the exhaustion, social isolation, and financial stressors that accompany caring for a child with a disability (Strunk 2010). However, in the United States services are provided through a patchwork of loosely connected organizations, including medical institutions, schools, informal support groups, and state and private agencies. Coordinating care across this complex landscape is a difficult undertaking (Foster et al. 2021; Lutenbacher et al. 2005), with families frequently encountering access barriers and delays, communication gaps between providers, and confusion about the criteria that qualify a child for services (Strickland et al. 2015; Cady and Belew 2017; Hasting, Lumeng and Clark 2014). Studies indicate that children with intellectual or developmental disabilities are less likely to receive needed community-based services, and report poorer quality health care experiences, than children with healthcare needs related to a primarily physical chronic condition (Cheak-Zamora and Thullen 2017; Graaf et al. 2021). Challenges obtaining needed services and experiences of social isolation can be particularly acute for families who are low-income, racially minoritized, and those whose primary language spoken at home is not English (Yu et al. 2015; Nguyen et al. 2016; Porterfield and McBride 2007; Rosen-Reynoso et al. 2016; Blanche et al. 2015).

Given how difficult it is to obtain and coordinate appropriate assistance, it is not surprising that parents whose child has an unexplained or unspecific intellectual or developmental condition might view genomic testing as a beacon of hope. Alongside the possibility of a causal explanation, new medical treatments, and insight into what can be expected in the future, families may envision etiologic knowledge as a compass that will guide them on their “therapeutic odyssey”—a search for help that often precedes genetic testing and almost inevitably continues long afterward (Brosco 2018). Thus, families often anticipate that genetic information will help them to legitimize their needs and to secure support services outside of the clinical setting. Providing empirical findings about these hopes, a recent study has shown that “some who received a positive sequencing result were able to leverage that result into direct benefits” (Childerhose et al. 2021). However, the anticipation that a genetic explanation will resolve the therapeutic odyssey may ultimately reinforce genomic medicine’s problem of “inflated promises” (Tabor and Goldenberg 2018), particularly given the lack of consensus about the role that genetic information should play in access to services.

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For children with intellectual and/or developmental differences, a *clinical* diagnosis is made by a clinician through assessment of anomalous neurological functioning across several areas – cognition, vision, motor coordination, hearing, speech, and behavior – and may or may not result in the assignment of a syndromic label such as Autism Spectrum Disorder. A *genetic* diagnosis is determined through genetic testing that indicates the molecular etiology of a child's phenotype. Depending on how much is known about the genetic variant(s) in question, a genetic diagnosis may also help to explain or predict developmental or functional characteristics. As with a clinical diagnosis, a genetic diagnosis may or may not result in the child's condition being associated with a named syndrome. Preliminary research has found that clinical diagnoses are useful to families as they search for services, although hopes that a genetic diagnosis will prove equally valuable have been met with mixed results (Childerhose et al. 2021; Lucas et al. 2022). More knowledge is needed about families' experiences outside the clinic after sequencing, in particular efforts to mobilize genetic information and how socioeconomic disadvantage shapes and constrains these efforts. This gap in our knowledge is particularly significant given genomics researchers' recent embrace of a paradigm of "inclusion and difference" (Epstein 2007), which has spurred recruitment in populations that have been historically underrepresented in biomedical research (see, for example, Amendola et al. 2018). However, more diverse participation has not resolved – and may even distract attention from – the social structures that shape how the benefits of genetic information are distributed (Fox 2020; Lee et al. 2022). It cannot be assumed that expanding enrollment of historically underrepresented in genomics research will lead to equitable benefit, particularly given existing inequities in access to both clinical and community-based services.

To understand the value of genomic sequencing, researchers must extend their scope of inquiry outside the clinic to encompass families' interactions with public and private agencies and service providers. However, studies of the utility of genomic sequencing have largely focused on clinical and psychosocial benefits (Lee et al. 2022; Kohler et al. 2017; Hayeems et al. 2021), with less attention on how genomic results mediate access to a broad range of therapeutic and educational services. Notably, a genetic diagnosis or etiology does not currently have a clearly defined role in determining eligibility for services. Rather, eligibility determinations draw primarily on clinical diagnoses and functional criteria (Williams 2017). For families of children with a suspected genetic condition, particularly those who depend on an array of publicly funded services, a key question remains: what happens when parents attempt to mobilize genetic information in their efforts to secure services for their child outside the clinic?

This paper reports findings from an ethnographic study with predominantly socioeconomically disadvantaged families enrolled in a multi-year genomics research project. Drawing on observations of clinic interactions and in-depth interviews, we examine parents' expectations of genomic sequencing and their efforts to use a genetic diagnosis to advocate for their child in negotiations with schools and social service agencies. We explore the current limits of rare genetic diagnoses' translatability from clinical to non-clinical service settings, the mismatch between parents' expectations and the eligibility criteria used by service providers, and the ways in which socioeconomic disadvantage and a fragmented service sector may further constrain the potential value of genetic information for families.

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Methods

Study Setting

The Program in Prenatal and Pediatric Genomic Sequencing (P^3EGS) is a clinical research study based at the University of California, San Francisco and a member of the Clinical Sequencing Evidence-Generating Research (CSER) Consortium. Studying the utility of exome sequencing (ES), P^3EGS aimed to enroll a high proportion of participants from medically underserved populations and/or groups historically underrepresented in genetics research. Between 2017 and 2021, 529 children with a suspected genetic condition – the majority with clinically observed intellectual and/or developmental differences – enrolled in the pediatric arm of the study. Families were referred to the genetics clinic from multiple healthcare institutions across California. To understand families' experiences undergoing ES, reactions to receiving positive, negative or uncertain findings, and experiences incorporating genetic information into daily life, we conducted clinic observations and longitudinal interviews.

The study was approved by the IRB at the University of California, San Francisco (protocol 17–23118).

Recruitment

Parents or legal guardians of children enrolled in P^3EGS were approached during study enrollment sessions at genetics clinics and asked for permission for a researcher to be present at the session and to be contacted about participating in interviews. Before return-of-results sessions, families who had agreed to be observed and contacted were purposively selected for interviewing, with a researcher observing the results return session and then inviting the families to participate in interviews. To explore a range of experiences before and after receiving a positive result, and given that most (73%) pediatric participants received a negative or inconclusive result, we oversampled for families with a positive result. Our purposive sampling strategy also considered demographic characteristics, including parents' self-reported racial or ethnic identity, rural/urban residence, language spoken at home, and health insurance coverage status, with the goal of creating a diverse sample with a high proportion of participants from communities that are socioeconomically disadvantaged and historically underrepresented in medical research. For the purposes of our study, we included families in the category of socioeconomically disadvantaged if they were low-income, had lower educational attainment, and/or had limited English literacy. Given the diversity of our study population, we estimated that enrolling approximately 30 families would enable us to approach "informational redundancy" (Sandelowski 1995). We contacted participants by phone to schedule interviews at the family's home, by phone, or via videoconference, depending on participants' preferences. Home interviews were discontinued in February 2020 due to the COVID-19 pandemic.

Data Collection

Interviews and observations were conducted as follows by the first and last authors, both experienced ethnographers, and two research assistants with training in qualitative research – one of whom is fluent in Spanish: 1) observation of study enrollment sessions; 2)

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observation of return-of-results sessions; 3) interview two weeks after results return; and 4) follow-up interview six months later. A semi-structured interview guide was used with questions for both interviews focusing on families' diagnostic journeys, expectations of ES, and the role of genetic information in healthcare decisions, family relations, and daily life. The 6-month follow-up interview also included questions about health-related decisions, family communication, service access, and daily activities that took place during the intervening months. All interviews were audio recorded, as were return-of-results sessions when permission could be obtained. Detailed fieldnotes were written after each clinic observation. All fieldnotes written by research assistants were reviewed and commented on by the two ethnographers.

Data Analysis

Interviews were professionally transcribed and Spanish-language interviews were first transcribed into Spanish and then translated into English by a bilingual transcriptionist. All transcripts were checked for accuracy and the Spanish-to-English translation was checked and edited as needed by the bilingual research associate. Transcripts and fieldnotes were uploaded to Dedoose (Version 9.0.17), a qualitative data management application. Six study team members participated in an inductive "flexible coding" and analysis process (Deterding and Waters 2021), which included iterative development and testing of 45 codes and sub-codes. After the team reached consensus that the codebook covered the full range of topics that were covered in the interview guide, as well as those that emerged spontaneously in interviews and observations, the codes were tested during a "blinded" coding process, in which two researchers separately apply codes to the same text and then discuss and reconcile any discrepancies in code application. Codes were then applied to all transcripts and fieldnotes by two team members.

Development of the findings presented in this paper began shortly after data collection started, with researchers conducting clinic observations noting that many families asked about the value of ES for access to community-based services. To further explore this emerging finding after data collection was complete, the team examined a sub-set of coded data pertaining to families' diagnostic journey, expectations of ES, and community-based services and supports. The team met frequently to develop and refine themes and resolve differences in interpretation. Themes developed from coded data not used for this analysis are presented elsewhere (e.g. Brown et al. 2022; Norstad et al. 2021; Outram et al. 2022).

Results

A total of 102 clinic observations (49 enrollment and 53 return-of-results sessions) and 61 interviews were conducted with 32 families. Approximately half of the interviews were conducted in Spanish. Mothers were more likely to participate than fathers (Table 1). Nearly all interviewed families were enrolled in Medicaid and half received a positive ES result (Table 2; in comparison, 27% of the overall pediatric cohort received a positive result). Below, we start with a narrative account of one family whose story illustrates themes identified in observations and interviews. We then provide an in-depth examination of two overarching themes: the first examining expectations of ES and drawing on insights from

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all participating families, regardless of ES result; and the second focusing on attempts to leverage a genetic diagnosis among families who received a positive or inconclusive result – the latter sometimes interpreted by the ordering clinician as diagnostic.

The Lopez Family[1]

Laura and Antonio Lopez have borrowed a car and traveled with their 6-year-old daughter Camila to the genetics clinic from their home approximately 90 miles from the medical center. Camila's condition includes developmental differences and low muscle tone and her pediatrician recommended that the family consider enrolling her in P^3EGS. They squeeze into a small exam room with members of the study team including a physician (Joan), genetic counselor (Priya) and ethnographer (Sam). Joan and Priya explain exome sequencing, emphasizing that it is unlikely to resolve all uncertainties about Camila's developmental and physical differences but that the results could inform her future clinical care. As they discuss the study's consent form, Laura is eager to explain the family's years-long effort to understand the health problems Camila has had since birth. As if in appeal, she says the child is having a hard time in school, particularly with speaking and writing, and that she will need more help than she is receiving in order to make it through life successfully.

Several months later, the family makes a second trip to the clinic to receive Camila's ES results. Joan explains that a rare genetic variant was detected and is likely to be the cause of Camila's symptoms. Laura asks if there is a treatment for Camila's condition. No, says Joan, but "the good news is overall, she's getting the help she needs…she's generally doing fine." Laura counters that Camila is not doing as well as she should be: "She's very behind in school."

During a subsequent interview with Sam, Laura explains what she expected from ES at the time of enrollment: "maybe if there was something that no one was trying to help me with and that she needed, that it would be a way to help my child in certain things that they couldn't– that they weren't trying to help her. So number one is school and other things, doctors, stuff like that…" She is relieved that the family is finally able to understand the cause of Camila's condition after such a long search: "I wasn't getting any answers with anybody else and any other doctor…I thank God that genetics gave me all this information, you know?" She presented the lab report to the school and requested that Camila's 30-minute special education sessions be expanded. The response was encouraging, and Laura is confident that the etiologic diagnosis will continue to open doors to assistance that will help her daughter to thrive.

At a follow-up interview six months later, Laura reports that her earlier optimism has not borne out. The school is "barely starting to give her a little bit more help… even though I took the genetics papers to the school, they didn't pay much attention to it… they don't understand a single thing of what genetics is saying..." This statement echoes the experiences of many parents we spoke with, who lamented that services were inadequate and were frustrated in their attempts to advocate for their child's needs. Despite this setback,

[1]Names and other identifying details have been changed to protect the family's privacy.

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Laura remains confident in the power of genetics: “without genetics you cannot help your child as much as you should, because genetics could tell you so many things.”

This account of one family’s experience brings into profile two overarching themes identified in interviews and observations and which we examine in more depth below: 1) Genetic diagnosis is a hoped-for solution; and 2) Families struggle to use genetic information beyond the clinic.

Genetic diagnosis is a hoped-for solution

During enrollment sessions, clinicians cautioned that not all families would learn the reason for their child’s condition and that a diagnosis was unlikely to result in a cure. Rather, they explained, genetic information could help guide decisions about referrals to specialty care and clinical management. Steered away from hopes of a medical cure, parents nonetheless often voiced a desire for a “solution.” Solutions were not conceived as a radical turnaround in a child’s features or symptoms. Rather, as in the Lopez family’s case, expectations coalesced around a broadly conceived expansion of information, understanding, and assistance that would enable families to improve the quality of their child’s day-to-day life.

Parents’ conceptualization of care extended beyond clinical services into the home and community. Many families regularly interacted with special education teachers, occupational therapists, social workers, and other service providers with whom they sought a shared understanding of a child’s needs. Etiologic information, parents hoped, would improve this mutual understanding and reduce the likelihood of inappropriate care. One parent voiced this hope when asked about her expectations of ES:

> Just to make sure that my daughter had very good, you know, well-being and she was raised properly in all that she needs or her necessities were taken care of. Because, you know…you might take your child somewhere or drop your child off somewhere and something goes wrong and you think that it’s somebody else’s fault [when the event may actually have been prompted by a misunderstanding of her child’s condition] … So that’s [learning more about her child’s condition through ES] just a part of being a responsible parent…
>
> (ID223, Positive Result, English spoken at home)

Parental responsibility for choosing appropriate childcare outside the home was in tension with the frequent difficulties they encountered obtaining adequate services. It is not surprising, then, that many families entered the study hoping that a genetic diagnosis could directly assist in their attempts to qualify for services that had previously been unavailable to them. For example, while waiting in an exam room before an enrollment session, a parent asked an ethnographer whether genetic information could help her family qualify for IHSS [In-Home Supportive Services], a state program that pays for home-based care for low-income people with disabilities. Their previous application had been denied, she explained, and she and her husband desperately needed help caring for their son.[2]

[2]The ethnographer responded by recommending that the parent direct the question to the geneticist and/or genetic counselor.

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In interviews, similarly, families anticipated that a genetic diagnosis could help them in their ongoing search for support services that would remedy a history of inadequate care and enable their child to thrive:

> It took her until she was like eight before she even saw a speech therapist…[she] kind of fell through the cracks, you know? […] So people can understand, like they'll say, okay, yes, she does need extra help and it's not just going to be relied on your shoulders, that there's actually help out there for her to help her get where she needs to be the best she can be.
>
> (ID282, Positive result, English spoken at home)

A timely diagnosis might, parents hoped, prevent children from "falling through the cracks" by validating their needs as much as possible. Nonetheless, using this information was not so straightforward.

Families struggle to use genetic information beyond the clinic

Families described a range of experiences as they attempted to leverage genetic information to obtain or augment community-based services. While some found test results to be useful for securing services, more often parents were frustrated that etiologic information did not translate into improvements in their child's day-to-day life. There were several circumstances that shaped the likelihood of parents making headway, starting with the timing and type of diagnosis.

An early and well-recognized genetic diagnosis opens doors—Although a very rare variant was the most common positive or inconclusive ES result among our participants, we also spoke with parents whose very young child did not have a previous clinical diagnosis and whose ES result was associated with a known genetic syndrome. These parents often reported success in finding structural supports. The mother of a two-year-old, for example, explained that presenting her child's genetic diagnosis of Wiedemann-Steiner syndrome to service providers immediately prompted new supports:

> Participant: He just turned three and he's already in Head Start, he goes to school three hours a day.
>
> Interviewer: Was the genetic testing important to get Head Start?
>
> Participant: Yes. The genetic testing gave us a diagnosis, where in turn the diagnosis lets us know what possible problems go along with his syndrome, so we're able to get a jumpstart on it and give him early treatment. Assistance, treatment, et cetera.
>
> (ID27, Positive result, English spoken at home)

A well-recognized genetic diagnosis also enabled some families to tap into a network of support groups and advocacy organizations that offered advice about care planning and negotiating with schools:

> I think what helps me is knowing that there are resources out there or like the support group that I found that I could ask what I could do when it comes time for

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> him to start school, if there's like a certain school that might help him out more…or what should I do if I don't agree with what the school is saying about him.
>
> (ID136, Inconclusive result reported by the physician as a "probable diagnosis" associated with a named syndrome, English spoken at home)

A genetic diagnosis is less helpful than anticipated—In contrast, parents facing prospects associated with an extremely rare diagnosis often reported that they could not locate or access online support groups for families with children like theirs. Further, while some families assumed that genetic information should be actionable in the context of therapeutic services, others grasped that service providers were primarily focused on specific behaviors and functional goals. The underlying etiologic picture suggested by a genetic diagnosis was less meaningful in this context, leaving the task of bridging these distinct diagnostic domains to parents:

> You know, it's so unusual, it's so rare, this condition, that – and therapists and other professionals…they have an area of expertise and that's what they focus on, and it's sort of up to us, as parents, to do our best to direct these things in a proper way.
>
> (ID101, Positive result, English spoken at home)

Indeed, we learned of numerous cases in which an etiologic diagnosis was not readily translatable to the assessment and treatment logics used by community-based service providers. Parents tried to fill these gaps themselves, yet their attempts to advocate for their child by explaining ES results often ended in communication breakdowns that were exacerbated by language differences. One parent, for example, struggled to explain her child's genetic diagnosis when applying by phone for assistance from IHSS. The screening questions focused on functional "symptoms," such as whether and where the child needed a walker, whereas the ES result indicated phenotypic and behavioral traits that the parent struggled to put into words that would be legible to the IHSS screener. Hoping that the lab report could help to resolve the impasse, she offered to fax it over:

> "Oh, no [the IHSS employee said], you have to do everything over the phone…I can't give you any fax number for you to send me any documents." I don't know if she didn't understand or I was unable to make myself clear…in the end, I decided to hang up and I didn't do anything. However, I would like to try again but I'm a little bit scared that the person who answers doesn't understand my daughter's illness or the problem she has.
>
> (ID268, Positive result, Spanish spoken at home)

Notably, this phone conversation took place via an interpreter. Parents with limited English literacy struggled to explain their child's ES findings to service providers, which compounded the challenge of finding support for rare genetic conditions. Communication challenges were also encountered by English-speaking parents, including Laura (see the Lopez family's story above), who made a plea for help from the clinic to strengthen her case with the school district:

> …even though I took the genetics papers to the school, they didn't pay much attention to it… So to me, it would be so much help for Genetics, if they're able to

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> come to see, whoever can come, and explain to special ed, you know, or even the school district how important it is for this child to get that help.
>
> (ID77, Positive result, Spanish spoken at home)

Ambivalence about the value of a genetic diagnosis—A protracted struggle to obtain services or accommodations for a child can lead families to conclude preemptively that a genetic diagnosis is of limited value, even if it offers the most definitive explanation to date for a child's condition. This was the case for a family whose ES results provided an etiologic explanation and clearly indicated the type of accommodation the child needed in order to remain in school. However, the parent chose not to share the ES result with the school because previous efforts to explain her daughter's condition had not resulted in meaningful change:

> …every year I have to go through this with the teachers. They think she's playing around… Honestly. I've had the doctors already write letters. I've given them to the schools. I've given them to the teachers and it's still problem.
>
> (ID303, Positive result, Spanish and English spoken at home)

Other parents persisted in their effort to use their child's diagnosis in negotiations with schools and social service agencies, even after repeated frustrations: "I have fought for so many things and I haven't succeeded," a parent told us after multiple attempts to persuade her daughter's school to discuss the ES result (ID231, Positive result, Spanish spoken at home). Despite her frustration, this parent planned to seek help from a local non-profit advocacy organization. She and others remained hopeful for a future in which a rare genetic diagnosis would prove useful to themselves and their child.

Discussion

Focusing primarily on the experiences of socioeconomically disadvantaged families, our findings demonstrate that the anticipated value of a genetic diagnosis extends beyond the clinic to encompass a web of community-based educational and therapeutic services that provide essential support to socioeconomically disadvantaged families. Many parents hoped that genetic information would help them to advocate for their child's needs as they navigated this complex landscape, contending with language barriers and opaque assessment criteria. Importantly, parents' search for understanding of their child's condition was entwined with an ongoing need for assistance in their effort to provide a good life for their child. For some, a genetic diagnosis opened doors to new forms of support, as has been reported in recent research.[21] More often, however, parents were unable to leverage a genetic diagnosis in their attempt to obtain services that they felt were essential for their child to thrive.

To some extent, families' ability to convert a genetic diagnosis into an effective advocacy tool was a function of "cultural health capital," or the skills, knowledge and interactional styles used to influence interactions with service providers. These skills include "linguistic facility, a proactive attitude toward accumulating knowledge, the ability to understand and use biomedical information, and an instrumental approach to disease management" (Shim

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2010). As we have written about in the context of exchanges between clinicians and parents about pediatric exome sequencing among disadvantaged populations (Outram, Brown and Ackerman 2022), there can be social and moral value experienced by parents in their choice or opportunity to seek more genetic answers. Nonetheless, providers' interactions with families who cannot enact these behaviors and skills – including many of the non-English speaking families in our study – may inadvertently reproduce upstream social inequalities (Dubbin, Chang and Shim 2013). In other words, fraught interactions with school officials, therapists, and other practitioners become a site where power differences and inequitable outcomes are reinforced, perhaps undermining any empowerment felt initially in the clinic.

It is important to emphasize, however, that even well-resourced parents struggle to translate genetic information outside the clinic, where eligibility criteria and service provision are based more on behavioral and clinical assessment than etiology. This is particularly true in the case of very rare genetic diagnoses, which may be of limited value for assessing a child's educational and therapeutic needs unless it points to a well-characterized syndrome or developmental trajectory. The mismatch between parents' expectations for a genetic diagnosis, and the criteria that schools and other service providers use to make decisions, was rarely if ever discussed with families during observed clinical interactions. As a result, families may over-estimate the current value of a rare genetic diagnosis, potentially resulting in a disproportionate burden on socioeconomically disadvantaged families who can little afford misspent time and resources.

Our findings also suggest the need for expanded conceptions of utility. In addition to accounting for the usefulness of genetic information for reassuring families and identifying potential treatments (Hayeems et al. 2021; Kohler, Turbitt, and Biesecker 2017; Mollison et al. 2020), it is critical to discern the impact of genetic information on access to supports outside the clinic and whether these benefits are equitably distributed (Smith et al. 2021). This is an important question for socioeconomically disadvantaged families, not only because they depend on an array of publicly funded services but also because their access to clinical genomics is often confined to research studies due to limited insurance coverage. Given the lack of clarity about the post-study obligations of clinical genomics researchers (Halley, Ashley, and Tabor 2022), and the absence of long-term follow up in many genomics studies, families who are only able to access genomic testing through research may receive inadequate guidance on how to understand and make the best use of their child's genetic information beyond the clinic (Gutierrez et al. 2021). Recent research indicates that the perceived utility of genomic sequencing is lower among disadvantaged families, thereby confirming the importance of a deeper examination of how genetic information operates at intersection of equity and utility (Halley et al. 2022).

The primary limitation of our study is the lack of a comparative population comprised of socioeconomically advantaged families, which would have enabled us to discern similarities and differences in the experiences of more and less advantaged families undergoing genomic testing and seeking therapeutic services for their child with unexplained intellectual and/or developmental differences. Additional research is needed to explore whether socioeconomically advantaged families are more effective at leveraging a genetic diagnosis than disadvantaged families, even when service providers identify the diagnosis as having

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little to no impact on service eligibility. Additionally, the genetic information conveyed to families in our study was usually associated with a rare condition. It is possible that using genetic information to access community-based services is easier for families whose child has a less rare condition – particularly if the condition and its developmental and functional implications are already known to service providers. Finally, access to services among the families in our study was shaped by California legal mandates and school district policies. As a result, the transferability of our findings to other states and countries may be limited.

In conclusion, we found that some families experienced a disjuncture between their hopes for genomic sequencing and post-results encounters with public institutions with complex and opaque service allocation logics. The resulting frustration may have been particularly acute among families who lacked the resources and know-how to recruit a lawyer, physician or other expert to support them in their struggle to obtain services from a school district or other public institution. To ensure that clinical genomics does not reinforce existing structural inequities by disadvantaging those who are already underserved, families would benefit from dedicated care coordination assistance to help them navigate the complex service landscape and convey genetic information and its implications to schools and other service providers. Additionally, the implications of a genetic diagnosis are often unclear in the context of community-based service provision, and research is urgently needed on how to best integrate etiologic information with clinical and behavioral assessments for children with unexplained intellectual and/or developmental differences. More concerted efforts to assist families outside the clinic, and explicit information for families and service providers about how genetic information may be relevant for functional needs assessment, could help to ensure that genomics contributes to efforts to provide a consistently supportive and enriching environment for children with rare genetic conditions.

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Table 1.

Interview participants and language

Family member participating	2 weeks after results	6 months after results
Mother only *	23	23
Father only	3	4
Mother & Father	6	2
Interview language		
Spanish	14	10
Spanish & English	1	1
English	17	18
TOTAL	32	29

* One family caregiver is included in the “mother” category in order to protect her privacy.

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Table 2.

Children's exome sequencing results and parents' demographics

	Participant, n (%)*
Exome sequencing result	
Positive	16 (50%)
Inconclusive	7 (22%)
Negative	9 (28%)
Self-identified race or ethnicity*	
American Indian/Alaska Native	2 (6%) (Mother)
Asian	4 (13%) (Mother) 3 (9%) (Father)
Black or African American	3 (9%) (Mother) 3 (9%) (Father)
White/European American/Middle Eastern	7 (22%) (Mother) 7 (22%) (Father)
Hispanic/Latinx	20 (63%) (Mother) 15 (46%) (Father)
Health insurance	
Medi-Cal (Medicaid)	29 (91%)
Private/Employer	3 (9%)
Education**	
High school or less	19 (59%) (Mother) 11 (34%) (Father)
Some college	9 (28%) (Mother) 5 (16%) (Father)
Undergraduate degree or more	3 (9%) (Mother) 4 (12%) (Father)

* Some participants chose one or more race and ethnicity categories.

** Education data is missing for 1 mother and 12 fathers, therefore percentages may not represent the full study population.